

Figure legends

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Supplementary Table 1 Effect of gender on the distribution of SNPs in VKH syndrome and controls

SNP	Allele		Male(MAF)		Pc Value	OR (95% CI)	Female(MAF)		Pc Value	OR (95% CI)
			VKH	Controls			VKH	Controls		
rs77258390	A	Chinese Singaporean	8	3	6.4×10^{-4}	11.5(2.8-46.2)	4	4	NS	2.5(0.5-10.8)
		Non-Han Chinese	6	10	NS	1.3(0.4-4.1)	13	7	NS	2.7(0.9-7.6)
		Thai	2	0	-	-	8	2	NS	2.7(0.5-13.1)
		Korean	2	3	NS	2.2(0.3-14.4)	2	6	NS	0.9(0.2-4.7)
rs3762318	C	Chinese Singaporean	8	5	8.6×10^{-4}	6.8(2.0-22.3)	5	4	NS	3.2(0.8-13.2)
		Non-Han Chinese	6	12	NS	1.0(0.3-3.2)	15	7	0.02	3.3(1.2-9.2)
		Thai	3	0	NS	-	9	3	NS	2.0(0.5-7.9)
		Korean	2	3	NS	2.2(0.3-14.4)	2	6	NS	0.9(0.2-4.7)
rs78597810	C	Chinese Singaporean	8	3	6.4×10^{-4}	11.5(2.8-46.2)	4	4	NS	2.5(0.5-10.8)
		Non-Han Chinese	6	10	NS	1.3(0.4-4.1)	13	7	NS	2.7(0.9-7.6)
		Thai	2	0	-	-	8	3	NS	1.7(0.4-7.1)
		Korean	2	3	NS	2.2(0.3-14.4)	2	6	NS	0.9(0.2-4.7)
rs12568393	A	Chinese Singaporean	8	3	6.4×10^{-4}	11.5(2.8-46.2)	4	4	NS	2.5(0.5-10.8)
		Non-Han Chinese	6	10	NS	1.3(0.4-4.1)	13	6	0.04	3.2(1.1-9.5)
		Thai	2	0	-	-	8	3	NS	1.7(0.4-7.0)
		Korean	2	3	NS	2.2(0.3-14.4)	2	6	NS	0.9(0.2-4.7)
rs12561798	C	Chinese Singaporean	8	3	6.4×10^{-4}	11.5(2.8-46.2)	4	4	NS	2.5(0.5-10.8)
		Non-Han Chinese	6	10	NS	1.3(0.4-4.1)	13	7	NS	2.7(0.9-7.6)
		Thai	2	0	-	-	8	3	NS	1.7(0.4-7.1)
		Korean	2	3	NS	2.2(0.3-14.4)	2	6	NS	0.9(0.2-4.7)

rs76436269	A	Chinese Singaporean	8	3	6.4×10^{-4}	11.5(2.8-46.2)	4	4	NS	2.5(0.5-10.8)
		Non-Han Chinese	6	10	NS	1.3(0.4-4.1)	13	7	NS	2.7(0.9-7.6)
		Thai	2	0	-	-	8	3	NS	1.7(0.4-7.1))
		Korean	2	3	NS	2.2(0.3-14.4)	2	6	NS	0.9(0.2-4.7)
rs78377598	T	Chinese Singaporean	8	5	8.6×10^{-4}	6.8(2.0-22.3)	4	5	NS	1.9(0.5-7.9)
		Non-Han Chinese	6	10	NS	1.3(0.4-4.1)	14	7	0.04	3.0(1.1-8.4)
		Thai	2	0	-	-	8	3	NS	1.7(0.4-7.1)
		Korean	2	3	NS	2.2(0.3-14.4)	2	6	NS	0.9(0.2-4.7)
rs117633859	G	Chinese Singaporean	7	5	2.0×10^{-3}	6.6(1.9-22.7)	5	6	NS	2.0(0.5-7.3)
		Non-Han Chinese	6	10	NS	1.2(0.4-3.8)	13	5	0.02	4.2(1.3-13.0)
		Thai	2	0	NS	-	8	3	NS	1.7(0.4-6.9)
		Korean	2	2	NS	3.2(0.4-24.6)	3	8	NS	1.0(0.2-4.0)
rs1884444	G	Chinese Singaporean	13	35	NS	1.4(0.6-3.1)	15	22	NS	2.2(0.9-5.4)
		Non-Han Chinese	15	29	NS	1.2(0.5-2.8)	28	20	0.02	2.8(1.2-6.2)
		Thai	25	2	NS	2.3(0.7-11.5)	34	18	NS	1.3(0.6-2.7)
		Korean	7	35	NS	0.5(0.2-1.3)	17	45	NS	1.0(0.5-2.1)
rs10995276	A	Chinese Singaporean	4	11	NS	1.2(0.4-4.3)	4	7	NS	1.3(0.4-5.1)
		Non-Han Chinese	3	5	NS	1.3(0.3-5.8-)	13	12	NS	1.3(0.5-3.2)
		Thai	13	1	NS	2.2(0.3-18.5)	14	4	NS	2.5(0.7-8.0)
		Korean	1	6	NS	0.5(0.06-4.5)	10	23	NS	1.3(0.5-2.9)
rs442309	T	Chinese Singaporean	7	24	NS	1.0(0.4-2.5)	6	18	NS	0.7(0.2-2.0)
		Non-Han Chinese	6	18	NS	0.6(0.2-1.8)	18	19	NS	1.2(0.5-2.8)
		Thai	30	2	NS	3.1(0.6-15.2)	26	10	NS	1.9(0.8-4.5)

		Korean	10	25	NS	1.5(0.6-3.8)	17	41	NS	1.2(0.6-2.5)
rs224048	T	Chinese Singaporean	7	24	NS	1.0(0.4-2.5)	6	18	NS	0.7(0.2-2.0)
		Non-Han Chinese	6	17	NS	0.7(0.2-1.9)	19	19	NS	1.3(0.6-3.0)
		Thai	30	2	NS	3.1(0.6-15.2)	26	10	NS	1.9(0.8-4.5)
		Korean	10	25	NS	1.5(0.6-3.8)	17	41	NS	1.2(0.6-2.5)
rs224052	A	Chinese Singaporean	7	24	NS	1.0(0.4-2.5)	6	18	NS	0.7(0.2-2.0)
		Non-Han Chinese	6	17	NS	0.7(0.2-1.9)	18	19	NS	1.2(0.5-2.8)
		Thai	30	2	NS	3.1(0.6-15.2)	26	10	NS	1.9(0.8-4.5)
		Korean	10	25	NS	1.5(0.6-3.8)	17	41	NS	1.2(0.6-2.5)
rs224057	A	Chinese Singaporean	7	25	NS	0.9(0.4-2.4)	6	18	NS	0.7(0.2-2.0)
		Non-Han Chinese	6	17	NS	0.7(0.2-1.9)	18	19	NS	1.2(0.5-2.8)
		Thai	30	2	NS	3.1(0.6-15.2)	26	10	NS	1.9(0.8-4.5)
		Korean	10	25	NS	1.5(0.6-3.8)	17	41	NS	1.2(0.6-2.5)
rs224058	T	Chinese Singaporean	7	24	NS	1.0(0.4-2.5)	6	18	NS	0.7(0.2-2.0)
		Non-Han Chinese	6	18	NS	0.6(0.2-1.8)	18	19	NS	1.2(0.5-2.8)
		Thai	30	2	NS	3.1(0.6-15.2)	26	10	NS	1.9(0.8-4.5)
		Korean	10	25	NS	1.5(0.6-3.8)	17	41	NS	1.2(0.6-2.5)
rs10995307	C	Chinese Singaporean	7	31	NS	0.7(0.3-1.8)	7	18	NS	0.8(0.3-2.3)
		Non-Han Chinese	4	18	NS	0.3(0.1-1.2)	18	17	NS	1.4(0.6-3.3)
		Thai	28	3	NS	1.6(0.4-6.7)	25	14	NS	1.2(0.5-2.6)
		Korean	9	24	NS	1.3(0.5-3.5)	16	37	NS	1.3(0.6-2.7)

Pc value: P value after Bonferroni correction; OR: odds ratio for minor allele; 95% CI: 95% confidence intervals.

Supplementary Table 2 Statistical power value of SNP association analysis in VKH syndrome patient groups from different Asian populations

Chr	SNP	Allele	Stage	MAF (Case/control)	OR	Power
1	rs77258390	A	Non-Han Chinese	25.0/14.2	2.0	0.46
	rs3762318	C		27.6/15.8	2.0	0.48
	rs78597810	C		25.7/14.2	2.0	0.46
	rs12568393	A		25.0/13.8	2.0	0.45
	rs12561798	C		25.0/14.2	2.0	0.46
	rs76436269	A		25.0/14.2	2.0	0.46
	rs78377598	T		26.3/14.2	2.1	0.51
	rs117633859	G		27.1/13.9	2.3	0.61
	rs1884444	G		56.6/45.8	1.5	0.28
1	rs77258390	A	Thai	6.2/3.0	2.1	0.17
	rs3762318	C		7.4/4.5	1.6	0.11
	rs78597810	C		6.2/4.5	1.3	0.07
	rs12568393	A		6.2/4.5	1.3	0.07
	rs12561798	C		6.2/4.5	1.3	0.07
	rs76436269	A		6.2/4.5	1.3	0.07
	rs78377598	T		6.2/4.5	1.3	0.07
	rs117633859	G		6.3/4.7	1.4	0.08
	rs1884444	G		36.4/30.3	1.3	0.13
1	rs77258390	A	Korean	5.9/4.5	1.2	0.06
	rs3762318	C		5.9/4.5	1.2	0.06
	rs78597810	C		5.9/4.5	1.2	0.06
	rs12568393	A		5.9/4.5	1.2	0.06
	rs12561798	C		5.9/4.5	1.2	0.06
	rs76436269	A		5.9/4.5	1.2	0.06
	rs78377598	T		5.9/4.5	1.2	0.06
	rs117633859	G		7.4/5.2	1.5	0.11
	rs1884444	G		35.3/40.0	0.8	0.12
10	rs10995276	A	Chinese Singaporean	12.5/9.6	1.3	0.09
	rs442309	T		20.3/22.3	0.8	0.09
	rs224048	T		20.3/22.3	0.8	0.09
	rs224052	A		20.3/22.3	0.8	0.09
	rs224057	A		20.3/22.3	0.8	0.09
	rs224058	T		20.3/22.3	0.8	0.09
	rs10995307	C		21.9/26.1	0.7	0.18
10	rs10995276	A	Non-Han Chinese	21.1/14.2	1.6	0.23
	rs442309	T		31.6/30.8	1.0	0.05
	rs224048	T		31.6/30.5	1.0	0.05
	rs224052	A		31.6/30.5	1.0	0.05
	rs224057	A		31.6/30.5	1.0	0.05

	rs224058	T		31.6/30.8	1.0	0.05
	rs10995307	C		30.3/30.2	1.0	0.05
10	rs10995276	A	Korean	16.2/14.5	1.1	0.06
	rs442309	T		39.7/33.0	1.4	0.21
	rs224048	T		39.7/33.0	1.4	0.21
	rs224052	A		39.7/33.0	1.4	0.21
	rs224057	A		39.7/33.0	1.4	0.21
	rs224058	T		39.7/33.0	1.4	0.21
	rs10995307	C		36.8/30.5	1.3	0.14

Power: We examined the power calculation using the Quanto program and assumed that the prevalence of VKH syndrome in Asian population is 0.001% according to the literature.

Supplementary Table 3 Allele Frequenc of IL23R-C1orf141 and ADO-ZNF365-EGR2 SNPs in different ethnic groups

	Alleles	Ethnic	AF
rs77258390	A	CEU	2.5
		CHB	10.8
		JPT	10.8
rs3762318	C	CEU	19.5
		CHB	20.7
		JPT	7.0
rs78597810	C	CEU	2.5
		CHB	10.8
		JPT	10.8
rs12568393	A	CEU	3.1
		CHB	20.7
		JPT	6.5
rs12561798	C	CEU	2.5
		CHB	10.8
		JPT	10.8
rs76436269	A	CEU	2.5
		CHB	10.0
		JPT	10.0
rs78377598	T	CEU	2.5
		CHB	11.7
		JPT	11.7
rs117633859	G	CEU	0
		CHB	10.8
		JPT	10.8
rs1884444	G	CEU	49.1
		CHB	45.1
		JPT	37.8
rs10995276	A	CEU	37.2
		CHB	8.1
		JPT	14.7
rs442309	T	CEU	43.3
		CHB	18.2
		JPT	26.7
rs224048	T	CEU	47.5
		CHB	27.5
		JPT	27.5
rs224052	A	CEU	49.2
		CHB	17.8
		JPT	31.1
rs224057	A	CEU	49.2
		CHB	17.8
		JPT	31.8
rs224058	T	CEU	54.0
		CHB	17.1
		JPT	30.8
rs10995307	C	CEU	49.6
		CHB	11.1
		JPT	32.0

CEU: Utah residents with Northern and Western European ancestry from the CEPH collection;
 CHB: Han Chinese in Beijing, China; JPT: Japanese in Tokyo, Japan.